

FEATURE ARTICLE

Genetic Support Groups in the Delivery of Comprehensive Genetic Services

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Summary

This research sought information about the services provided by genetic support groups, their members' experiences in obtaining genetic and related services, and members' recommendations for improving services. Results from a survey of 43 directors of genetic support groups showed that these organizations not only provide their members with a wide range of informational and supportive services but also address the need for education of both the public and health professionals about genetic disorders. A second survey of 931 members of genetic support groups found that, although they obtained genetic information from a variety of professional and informal sources, many of them experienced barriers to obtaining sufficient genetic information. Respondents called for professionals to improve their interpersonal skills in working with clients and to assist families in obtaining a wider variety of services. On the basis of these findings, a service model and priorities are proposed to bring together genetic specialists, community professionals, and genetic support groups for the delivery of comprehensive services to individuals and families with genetic disorders.

Introduction

With the rapid development of genetic support groups, health professionals increasingly are being asked to address the gaps in services identified by these voluntary organizations. Consumers have noted particular service deficiencies in financial counseling services, professional counseling services for emotional concerns, basic medical care, pastoral counseling, physical and occupational therapy, and speech therapy (Black and Weiss 1988). Genetic clinics encounter an uphill battle as they struggle to address these needs at the same time that they face both an increasing demand for their limited professional resources and increasing constraints

on funding for their services (Bernhardt et al. 1986). More important, the range of service needs extends beyond the genetic programs' generally more narrow mandates: to provide (1) diagnoses and prognoses, (2) limited, specialized medical management, and (3) genetic counseling.

These considerations underscore the importance of looking beyond formal genetic services to identify professional and lay community resources that genetic clients and their families may turn to for genetic information, counseling, and help in coping with the multiple ramifications of genetic disorders (Schild and Black 1984). Genetic support groups may be one such lay resource (Black and Weiss 1988), supplying both education about the genetic aspects of the disorder and help with adjusting to the personal or emotional meanings of the disorder. Groups also may provide help with numerous other genetic concerns that can arise among families and in reproductive decision making.

Received January 26, 1989; revision received May 30, 1989.

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0002-9297/89/4504-0021\$02.00



Although the preceding review suggests that more attention should be given to the comprehensive needs of genetic clients and that genetic support groups offer important possibilities for assisting clients, the available evidence remains quite limited and inadequate to suggest specific programmatic directions for enhancing current services. Systematic studies are needed that provide information about at least three areas: (1) factors that impede optimum access to and utilization of genetic counseling services, (2) major gaps in the professional community services needed by genetic clients and their families in order to cope effectively with the many ramifications of genetic disorders, and (3) services provided by genetic support groups for both specifically genetic as well as related concerns.

This paper reports the findings from two studies that contribute data, pertinent to these three areas, from large, national samples of directors and members of genetic support groups. Turning to the support groups for assistance builds on the developing alliance between genetic and community health professionals and the groups (Weiss et al. 1986) and offers a unique opportunity to obtain a consumers' perspective. While data obtained from directors and members of support groups will not be representative of all past or potential genetic counseling clients, the findings of Black and Weiss (1988) indicate that members of these groups do have certain criticisms about the delivery of genetic and related services and that they may themselves be providing some help with genetic concerns as part of their support-group activities. Thus, directors and members of genetic support groups represent a group of past and potential genetic counseling clients who are likely not only to have given some thought to the delivery of genetic services but may also have provided some level of services themselves. As a result, their experiences and suggestions provide an important and unique perspective to consider in developing a more comprehensive model of service delivery.

Material and Methods

Two groups of potential respondents were identified and contacted for participation in the surveys. For the first group (hereafter referred to as the directors' survey), questionnaires were mailed to the directors/executives of all service organizations listed, as of January 1988, on the directory of genetic organizations that is maintained by the National Center for Education in Maternal and Child Health. After those organizations that performed only fund-raising or research activities

were excluded, 76 genetic support groups remained for investigation.

The second survey (hereafter referred to as the members' survey) was sent to the members of service organizations who were affiliated with the Alliance of Genetic Support Groups as of January 1988. (The Alliance of Genetic Support Groups, formed in 1985, is a national network of voluntary genetic organizations. It is dedicated to fostering a partnership among consumers and professionals in order to enhance education and service for and to represent the needs of individuals and families affected by genetic disorders.) Fifteen (62.5%) of 24 of the groups then in the alliance agreed to participate in the survey of their members. (Of the nine organizations that declined, two cited inadequate administrative resources to handle a mailing, one cited a policy of not surveying its members, and six did not respond to the project's invitation.) In order to reach as broad a sample of members as possible while remaining within a limited project budget, a sampling plan was developed that allowed mailings to all parties on the mailing lists of the four organizations that had fewer than 250 members. For 10 larger groups, sampling fractions were developed to yield mailings to a randomly selected sample of 250 members (percentages of the 10 mailing lists sampled in this fashion ranged from 16% to 100%). One organization was unable to mail out questionnaires but did participate by distributing the surveys to all of the 200 families who attended the organization's annual meeting. In order to insure confidentiality of the organizations' lists, prepared envelopes were sent to each group for its own distribution. (See Appendix for additional details on the organizations that participated.)

Unless otherwise noted in the report of results, questions in the surveys asked respondents to choose among a predetermined set of response choices. Categories provided in tables 1–4 and other such responses referred to in the text closely follow the actual wording of items in the questionnaires.

Sections of the members' questionnaire that will be discussed in this paper concern (1) demographic characteristics of the respondents, (2) the bases for their connection to the genetic disorder represented by the organization, (3) the nature of their associations with the genetic organization, (4) their sources of genetic information, (5) factors that might have made it difficult for them to obtain desired genetic information, (6) the extent to which they were satisfied with the manner in which professionals had provided genetic information and related services, (7) the extent to which other services related to the genetic disorder had been needed



and received, and (8) recommendations for enhancing the training of genetic and other health professionals who serve patients and families with genetic disorders.

Also reported on are those sections of the directors' questionnaire that sought information on (1) demographic characteristics, (2) the nature of respondents' positions in the organizations, (3) the structure and size of the organization, (4) the general types of services provided, and (5) services related specifically to the genetic nature of the disorders.

Written comments also were encouraged at many points in the questionnaires to obtain elaborations of respondents' views. These comments were coded for major themes by one of the authors (R.B.B.).

Results

A total of 43 questionnaires were returned by the directors, for an overall response rate of 57%. Respondents were either directors of national organizations (34 [79%]) or international organizations (9 [21%]), with many of the groups also sponsoring local, state, or regional chapters. Comparison of membership sizes is complicated by this diversity in the organizational structures of the groups. In addition, some groups identified only affected individuals and families as members, while others listed all interested persons on their national mailing lists. Membership estimates thus ranged from fewer than 100 to 23,000, although the median size of 575 provides probably the best estimate of the most common size.

Respondents to the directors' survey occupied positions which they most frequently described as executive director, president, or other type of administrator. The structure of their positions also mirrored the organizational diversity, with 17 (39.5%) reporting that their position was full-time and paid; 4 (9.3%) part-time and paid; 10 (23.3%) full-time and volunteer; and 12 (27.9%) part-time and volunteer. The majority were female (35 [81.4%]), Caucasian (38 [88.4%]), and had completed at least some college training (38 [88.4%]); with 20 (46.5%) having completed graduate or professional training. A majority (32 [74.4%]) also reported a personal connection to the genetic disorder represented by their group, with parents of children with the disorder constituting the largest subgroup (18 [42%]).

The overall rate of return for the members' survey was 33% ($N = 931$). However, return rates varied considerably between the groups, ranging from virtually no returns from the sickle cell groups to 60% for the

National Mucopolysaccharidoses Society. (See Appendix for additional details.)

The 931 respondents to the members' survey were largely female (726 [78%]), Caucasian (873 [94%]), currently married (721 [78%]), working outside the home (571 [63%]), and had at least partial college training (583 [63%]). Average age of respondents was 40 years, with the majority between 29 and 51 years of age, although ages did range from 11 to 86 years. Five affected young people, 18 years of age or younger, completed questionnaires.

As might be expected from both the nature of the disorders and higher responses rates of certain of the groups (see Appendix), the majority of the member respondents (665 [74%]) were parents of affected children. However, close to one-third (299 [32%]) had the disorder themselves, with 95 (10%) reporting that both they and one or more of their children had a genetic disorder. Overall, 774 (83%) either were parents of affected children and/or had the disorders themselves.

For the most part, member respondents said their only involvement in the genetic organization was their sign-up for the mailing list (636 [68%]), although 176 (19%) indicated current or past participation in an interpersonal support group.

Services Provided by Genetic Support Groups

As shown in table 1, the directors described a wide variety of general services and activities conducted by their organizations. Education of members about the medical facts of the disorders emerged as the most frequent function, with education of health professionals and the public also cited as frequent activities. Self-help or peer-counseling services were offered by almost all of the groups, with the majority also providing some additional assistance in linking members to various community services and advocating for improved services.

Table 2 summarizes the types of services that the directors identified as pertaining specifically to the genetic nature of the disorders. Again, educational efforts are the most frequent service, with help in adjusting to the personal or emotional impact of the genetic disorder also a major focus of their efforts. The family nature of genetic diagnoses is reflected in the assistance provided both for discussing genetic issues with children and other relatives and in reproductive decision making.

Directors also were asked to describe any particularly helpful or creative services or activities performed by their organizations. The rich variety of programs reported in their written comments and in the brochures

**Table 1****General Services and Activities of Genetic Support Groups (N = 43)**

SERVICE/ACTIVITY	FREQUENCY (%)		
	Often	Sometimes	Never/Don't Know
Education of members about medical care, treatment, research for disorder	93	7	0
Education of health professionals about impact of disorder	79	21	0
Public education about disorder	70	28	2
Self help/peer counseling	58	30	12
Fundraising for medical/genetic research	53	14	33
Efforts to link members with community services	46	35	19
Fundraising for services	46	19	35
Advocacy efforts to improve services for people affected by disorder	30	49	21

attests to the diverse and innovative programming sponsored by these groups. Newsletters were cited most frequently as a vehicle for contact and information sharing, with many of the groups also developing specific informational materials about the disorders, for use by families or professionals.

At least 13 (30%) of the directors highlighted the benefits of their organizations' annual conferences. Held on a national or regional basis, these meetings exemplify the groups' dual commitments to education and services (see table 1) by bringing together affected individuals, families, and professionals for mutual education and support. Formal sessions usually include both professional presentations about recent scientific and medical developments and presentations by individu-

als and families about the personal impact of the disorder and about helpful coping strategies. Equally important are the opportunities provided for professionals and consumers to become acquainted during the many informal, recreational times offered at the meetings.

Members' Experiences and Service Needs

Respondents to the members' survey reported an average of two professional and two informal sources of genetic information (see table 3). With more people citing medical specialists other than geneticists as sources of genetic information, the prevalence of genetic counseling activities outside official genetic programs is evident.

Barriers to obtaining sufficient genetic information

Table 2**Support-Group Services Related Specifically to Genetic Nature of Disorders (N = 43)**

SERVICE/ACTIVITY	FREQUENCY (%)		
	Often	Sometimes	Never/Don't Know
Education about genetic aspects of disorder	81	14	5
Help adjusting to personal and emotional impact of genetic disorder	70	28	2
Help with concerns about informing extended family of genetic nature of disorder	42	35	23
Help with concerns about informing one's children about their reproductive risks	33	35	33
Help with reproductive decision making about genetic risks to future offspring	14	44	42

NOTE.—Rounding causes some percentages to not sum to 100%.

**Table 3****Members' Sources of Genetic Information (N = 931)**

Source	%
Professional:	
Other medical specialist	55
Geneticist (M.D./Ph.D.)	52
Pediatrician	32
Genetic counselor (non-M.D.)	18
Other health professional	13
Internist	12
No information from professional	12
Ob/Gyn	8
Other:	
Medical books/journals	64
Genetic support groups	53
Relatives and friends	18
Books for general public	17
Newspapers	16
No other sources	13
TV, radio	7

included limited professional knowledge about the disorder (713 [76.5%]), difficulties knowing where to go to get desired genetic information (540 [58%]), travel time (473 [51%]), financial costs (425 [46%]), and lack of flexibility in scheduling appointments (241 [26%]).

Respondents also were given a list of 20 possible ser-

vices that might be utilized in coping with the ramifications of a genetic disorder and were asked to indicate whether they had needed each service and, if so, whether the needed service had been received. Responses showed they had needed and used an average of five services (SD = 3.9) but had not received an average of three needed services (SD = 3.4). Services described by at least 10% of respondents as needed but not received are listed in table 4, along with the corresponding percentages of those who either obtained the same services or felt that the service categories did not apply to them.

Qualitative analysis of the members' written comments helped to confirm and elaborate responses to the fixed-choice items reported above. Difficulties in obtaining insurance was the most predominant theme in their remarks. Many reported that they or their children did not fit into clear categories of eligibility; those who had managed to obtain some sort of coverage often described long and frustrating appeal processes because of the rarity of their disorder and lack of knowledge on the part of Social Security or insurance officials. As one parent wrote, "financial aid and medical insurance has been the biggest example of frustration we have ever been involved in." Personal and family stresses generated by the multiple costs of the illnesses often were noted.

Table 4**Services Frequently Unavailable to Respondents (N = 931)**

Service	Needed But Not Received (%)	Need and Used (%)	Not Applicable (%)
Supportive counseling	35	33	32
Budget/financial planning	30	14	56
Applications	28	25	47
Help obtaining insurance	24	24	52
Recreational programs	21	16	63
Home care	20	19	61
Child care	18	20	62
Counseling by clergy	16	26	58
Respite care	16	20	64
Educational planning	15	48	37
Early intervention programs for infants/toddlers	14	33	53
Vocational/rehabilitation services	13	14	73
Occupational therapy	12	36	52
Physical therapy	11	51	38
General medical/dental care for affected individuals	11	77	13

NOTE.—Percentages were calculated as proportion of answered items. Nonresponses (not shown) varied from 4% to 8% across items.



The special burden of rare disorders appeared also in descriptions of struggles to get correct diagnoses, genetic information, and ongoing treatment.

The stresses generated by the work of managing chronic illnesses (Corbin and Strauss 1988) were evident in the anger and frustration expressed about the lack of adequate respite or home care services. The burden of finding and coordinating multiple professional service providers also often fell heavily on the affected person or parent. As the parent of a child with a mucopolysaccharide disorder wrote, "no program or help was offered to us. We . . . found and fought for every service my son has received. It's exhausting plus you don't know if you're doing the right thing without guidance." The genetic support groups often were cited as one of the family's main or only sources of useful information about the disorder and its management.

When support-group members were asked to make recommendations for enhancing the training of professionals about the needs of genetic clients, two major themes were evident in their suggestions. First, respondents called for professionals to improve their skills in conveying compassion and respect for clients, in communicating information clearly, and in listening empathically to families' concerns. Second, members called for professionals to become more knowledgeable about families' needs for a variety of services and to assist in referring their clients to appropriate programs. These themes are eloquently summarized in the suggestions of one respondent: "Listen to what your clients are saying, don't make snap judgments. Make yourself aware of parent support groups to put these families in touch with. If you don't know the answer to a question, have the guts to say so and offer to find someone who might know the correct answer. Make good use of the information and resources available to you. Be a little human. . . . A little kindness and caring goes a long way."

Discussion

The findings presented in the present paper suggest that many members of genetic support groups obtain their genetic information from a wide variety of professional and informal sources, but sizable proportions of these consumers are likely to have experienced barriers to receiving desired amounts of genetic information. These barriers include difficulties in finding knowledgeable professionals, lengthy travel time to major medical centers, and financial costs. The broad impact that genetic disorders have on patients and their families also often generates numerous service needs

that call for the expertise of professionals from a variety of disciplines. The difficulties reported in obtaining these related services underscore the importance of increased sharing of information and referrals between genetic and community professionals and genetic support groups.

The reports of both genetic support-group members and their leaders indicate that these organizations provide their members with a wide range of information about genetic disorders and associated medical problems, as well as supportive counseling services. These groups also are active in educating the public and health professionals about genetic disorders, in fund-raising for research and services, and in advocacy efforts to improve services.

Certainly caution must be exercised in generalizing these findings to all persons with genetic disorders, particularly those families from low-income and racial minority groups who were dramatically underrepresented in the responses. Nevertheless, these data do come from the first known attempts to survey broad, national samples of leaders and members of genetic support groups, and responses were obtained from a sizable number of persons both personally affected by and representing many different types of genetic disorders.

The consumer perspective reflected in these findings calls for a comprehensive model of genetic and related services, based on a close partnership between genetic specialists, community professionals, and genetic support groups. As shown in figure 1, no single member of this trio of service providers is sufficient for addressing the comprehensive needs of families; and maximum service quality will come only with the coordinated efforts of this trio. In addition, the need to address scarcities in technical expertise about and services for those with rare disorders calls for broad-based efforts that bring together genetic specialists, community professionals, and genetic organizations in state, regional, and national coalitions. Implicit in figure 1's visual representation of this model of collaborative service delivery are the following priorities:

Clinical Services

Education programs for all professionals who serve patients and families with genetic disorders should:

1. Enhance professionals' skills in communicating and counseling. Sensitivity to emotional reactions, respect for client's individual concerns and needs, and clarity of communication are essential elements of any helpful, professional intervention, but they be-

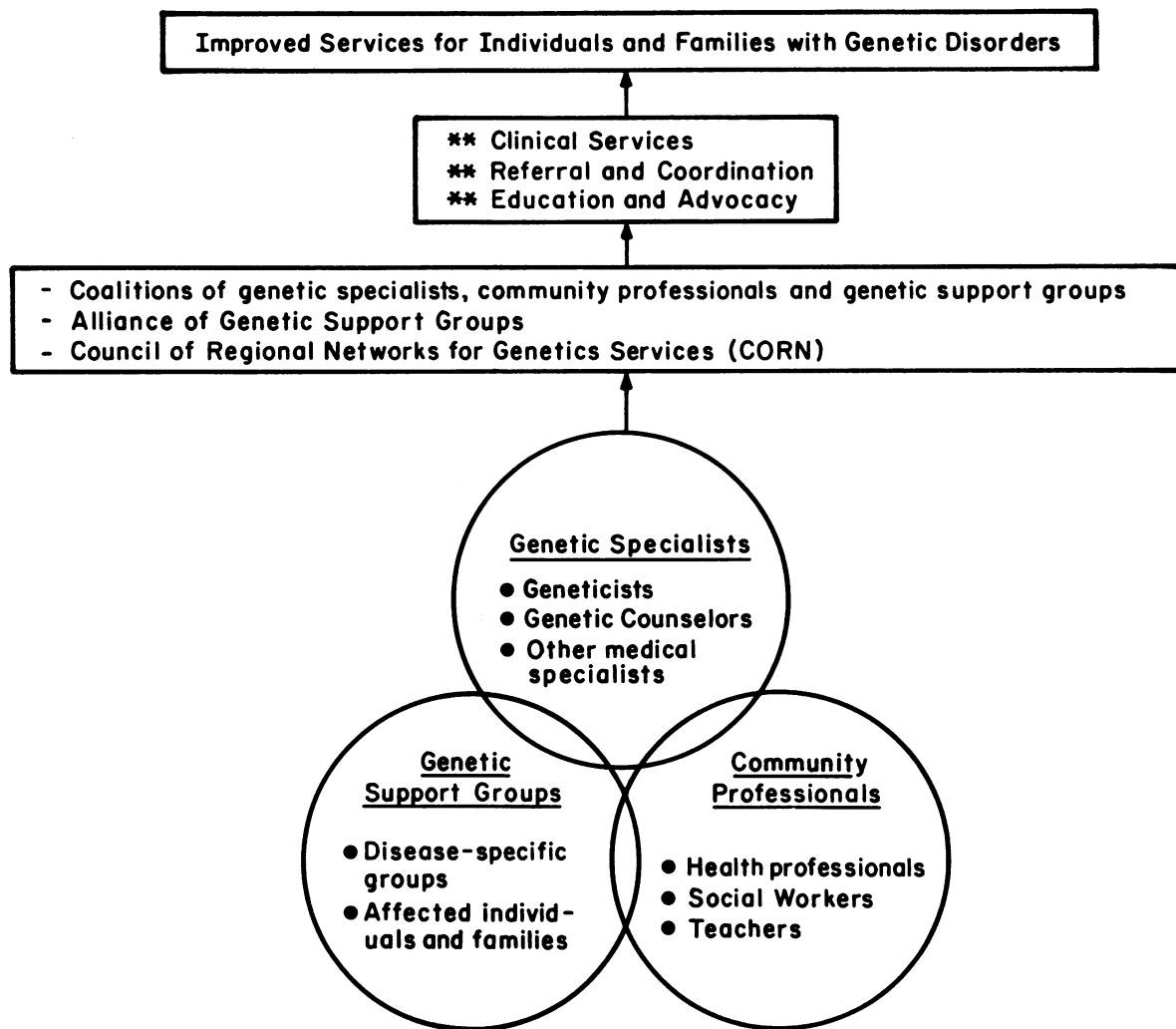


Figure 1 Model for delivery of comprehensive genetic services

come especially important for those individuals and families whose genetic disorders are rare, hard to diagnose and treat, and multifaceted in their impact.

2. Provide content on the comprehensive ramifications of genetic disorders and the sometimes subtle but personally significant variations in subtypes and phenotypes. Issues to consider include (a) progression or changes of symptoms over the life cycle and (b) the disorder's potential impact on cognitive, physical, social, emotional, vocational, recreational, and sexual functioning. Information should be included on the many types of diagnostic, therapeutic, educational, and support services pertinent to the needs of genetic patients and on the professional disciplines knowledgeable about each area.

3. Provide content on genetic support groups and the services they may offer, to both consumers and professionals, about specific disorders and their ramifications.

Referral and Coordination of Services

Genetic specialists, working in collaboration with community professionals and genetic support groups, should identify a comprehensive range of resources, in their community, state, and region, to which they can refer patients and families with particular genetic disorders.

In many instances, genetic support groups will be the critical agents for both collecting information about rare genetic disorders and referring families to appro-



Appendix

Organizations Participating in Members' Survey

Group	No. Returned (% ^a)
Association for Glycogen Storage Disease	75 (37)
Central MD Committee on Sickle Cell	1 (. . .)
Cystinosis Foundation	22 (20)
Ehlers Danlos National Foundation	120 (49)
Freeman-Sheldon Parent Support Group	12 (40)
G.I. Polyposis and Related Conditions: Newsletter	61 (25)
Hemophilia Foundation of Illinois	60 (24)
International Rett Syndrome Foundation	144 (59)
Long Island Sickle Cell Project	11 (5)
Lowe's Syndrome Association	23 (39)
National Foundation for Ectodermic Dysplasias	13 ^b (27)
National Marfan Foundation	108 (46)
National Mucopolysaccharides (MPS) Society	150 (60)
United Leukodystrophy Foundation	94 (38)
Wilson's Disease Association	37 (15)
Total	931 (33)

^a Calculated after ineligible and post office returns were subtracted from original number sent out.

^b Questionnaires distributed at annual conference.

priate geneticists and other specialized professionals. Therefore, all professionals with relevant areas of expertise should register with the appropriate genetic organizations and assist the groups in developing and maintaining up-to-date referral banks. The Alliance of Genetic Support Groups also can assist individual genetic organizations in developing their information

and referral capabilities and can help coordinate resources identified by different genetic groups and in different regions.

Education and Advocacy

Genetic specialists, genetic support groups, and involved community professionals should collaborate to educate the public, politicians, insurance executives, and government officials about the sequelae of rare genetic disorders and appropriate levels of treatment, education, rehabilitation, and disability/insurance coverage. The Alliance of Genetic Support Groups, regional genetics networks, and the Council of Regional Networks for Genetics Services (CORN) provide important structures on which to build such advocacy coalitions.

Acknowledgments

Support for this project was provided by March of Dimes — Birth Defects Foundation grant 14-74 and by Bureau of Maternal and Child Health and Resources Development, Department of Health and Human Services grant MCJ-009114-01-0.

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